The Impact of Online Images of Neurofibromatosis Type 1 on the Parents of Newly Diagnosed Children

Master’s Thesis

Presented to

The Faculty of the Graduate School of Arts and Sciences
Brandeis University
Graduate Program of Genetic Counseling
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In Partial Fulfillment of
the Requirements for the Degree

Master of Science
in
Genetic Counseling

by
Mary-Kathryn Cushing

May 2015
ACKNOWLEDGEMENTS

There are many individuals without whom this project would not have been possible, and to whom I am greatly indebted.

I would first like to thank the seven study participants from the Children’s Tumor Foundation NF Registry for sharing their experiences and insights.

I would like to express my deepest appreciation to my thesis advisor, Gretchen Schneider, MS, CGC, and committee members, Caroline McGowan, MS, CGC, and Sara Ellingwood, MS, CGC. Thank you for your continued support and encouragement, insightful advice, and dedication. Your enthusiasm and immense knowledge of this topic was invaluable.

I would like to thank Amy Spirito for her insightful feedback in designing the research tool for this project. I would also like to thank Liz Cross and Margarita Corral for their guidance regarding qualitative analysis and ATLAS.ti software.

Thank you to the Graduate School of Arts and Sciences and the Genetic Counseling Program for funding this project. Thank you to the Brandeis University Genetic Counseling program and faculty, especially Judith Tsipis and Missy Goldberg.

Finally, I extend my sincerest gratitude to my amazing classmates, family and friends for their unwavering love and support.
ABSTRACT

The Impact of Online Images of Neurofibromatosis Type 1 on the Parents of Newly Diagnosed Children

A thesis presented to the Graduate Program of Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Mary-Kathryn Cushing

Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder affecting approximately 1/3000 individuals. Due to the high de novo rate (50%) of the condition, many children are born to parents with no prior knowledge of NF1. Internet searches for NF1 images typically uncover pictures depicting the most severe end of the disease spectrum which do not accurately represent the disorder for most individuals. This qualitative study aimed to gather insights from parents of children with a new diagnosis of NF1, who have no prior knowledge of the disease and have viewed internet images of affected individuals. This study, involved semi-structured interviews with 7 mothers of children with a recent diagnosis of NF1 that focused on their diagnostic stories, first encounters with internet images of NF1, sharing of the pictures with other individuals and advice for future parents of children with NF1. Study participants ranged in age from 30 to 40 years and their children were between 2 and 3 years old. The participants felt that parents should be advocates for their children, with four mothers proactively seeking out NF1 information on the internet. Viewing online images caused every participants to have negative feelings such as worry or heartbreak. Overall, these mothers found that internet images
misrepresent the diagnosis; five felt the majority of the online images did not relate to their child and three felt the majority of online images did not relate to the NF1 population as a whole. The most memorable images for all participants were ones that represented very severe cases of NF1. All participants shared NF1 images with friends or family very selectively and viewed this as a personal choice, but felt their children should be introduced to the images with guidance at an age appropriate time. Advice for future parents of children with NF1 included seeking information from reputable sources and being aware that most people with NF1 are not as severely affected as the individuals depicted online. Insights from parents about their experiences with these can help healthcare providers, like genetic counselors, adequately address common concerns about NF1 images.
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INTRODUCTION

Neurofibromatosis type 1 (NF1) is one of the most common genetic conditions, occurring in approximately 1 in 3000 individuals. It is an autosomal dominant disorder characterized most commonly by neurofibromas, café au lait spots, skin-fold freckling, and Lisch nodules. NF1 has a 50% \textit{de novo} rate, therefore 50% of individuals diagnosed with NF1 have no family history of the condition (Radtke, 2007). NF1 is extremely variable, even among individuals in the same family, making the course of the disease unpredictable. However, the majority of affected individuals, two thirds of the NF1 population, are considered “mildly affected” (Radtke, 2007).

The clinical diagnostic criteria for NF1, established by the NIH Consensus Development Conference (1987), require the presence of two or more of the following traits to establish a diagnosis; six or more café au lait spots of a certain size, skin fold (axillary or inguinal) freckling, two or more neurofibromas or one plexiform neurofibroma, sphenoid or tibial lesions, two or more Lisch nodules, optic glioma or a family history of the condition. The most common symptoms of NF1 are generally mild and benign. These include café au lait spots, skin fold freckling and Lisch nodules, which occur in 99%, 90% and 95% of the NF1 population, respectively. The symptoms considered the most severe tend to be present less frequently; these include plexiform neurofibromas, scoliosis, malignant peripheral nerve sheath tumor, and optic glioma (Radtke, 2007). In all individuals with NF1, 25% have a plexiform neurofibroma, 10-25% have some degree of scoliosis and 4-13% have a malignant peripheral nerve sheath tumor (Radtke, 2007). Up to 15% of individuals with NF1 will have an optic glioma, however only a small proportion of these optic gliomas process and require treatment (Radtke, 2007).
In the setting of a genetic disorder with an unpredictable course, patients and families may seek out as much information as possible to gain a better understanding of the natural history of the condition. Many people supplement what they have learned from their healthcare providers with material from other sources, like the internet. A study that examined the information seeking behavior of parents of children with genetic conditions reported that 69% of these parents searched the internet for more information on their own, while an additional 14% had someone else search the internet for them (Roche, 2009). Parents who had conducted these internet searches, either on their own or with aid from someone else, were most frequently seeking guidance on what to expect in the clinic, how to prepare questions to ask, how to prepare answers to questions from healthcare professionals, and reviews of basic genetic concepts and terms. Parents classified internet material as supplemental, secondary to information gathered during clinic visits and those who used the internet as a resource reported feeling better informed about their child’s condition. However, parents also reported a need for guidance in assessing website reliability (Roche, 2009).

Despite the benefits to accessing medical information online, what patients or parents learn on the internet has the potential to become a barrier to the patient-provider relationship. Misinformation from unreliable sources on the internet can impede care when it causes patients to become skeptical of what they learn from their healthcare professionals (Johnson, 2014). In a 2010 study, Gallo et al. found that medical professionals reported frustration with the inaccuracies of what patients found on the internet and the time spent in clinic re-teaching and clarifying health related information. Yet even with clinicians’ concerns about their patients’ internet searching, it seems inevitable that many will use this resource to gain more information about their own health or that of their children.
For those seeking health-related material online, health literacy and readability of web resources are barriers to gathering and understanding reliable medical information from the internet. The extent of a person’s basic reading and numerical abilities are the major determinants of health literacy, and proficiency in both categories is required in the healthcare environment (Gutierrez, 2014). Therefore, an individual’s level of health literacy strongly correlates with how accurately they are able to interpret health information obtained online. The National Assessment of Adult Literacy reports that 36% of the population in the United States has basic or below basic health literacy (Kutner, 2006).

Although there are inequities in health literacy across the country, online information seeking behaviors do not tend to change across groups (Gutierrez, 2014). The average person in the United States reads at an 8th grade level, but not all internet information targets this level (Gutierrez, 2014; Mcinne, 2011). Health information websites discovered through popular search engines, including Google, ASK, Yahoo, Bing and AOL, with a “.gov” or “.nhs” address tend to have the highest readability, while “.edu” websites tend to have the lowest readability. The most frequent search results, such as entries from Wikipedia, tend to be the most difficult to read (Mcinne, 2011). In other words, the most accessible online material is not necessarily the most easily understood material for the average US healthcare consumer.

Patients or parents who conduct internet searches for health information will inevitably encounter photographs. Medical photography is an invaluable tool to both the clinician and patient when used appropriately. Accurate documentation of disease symptoms with photographs can give a concrete standard of comparison for both the patient as they grow, develop and age and for new patients presenting with similar symptoms. They also allow consistent tracking of medical outcomes and provide standards for other physicians to utilize (Katugampola, 2012;
Collard, 2011) When medical photographs are accessed by patients or families on the internet outside of a clinical setting and without physician oversight, the images patients view may or may not accurately depict the disease they are researching.

In the case of NF1, the web contains hundreds of medical photographs depicting the most severe end of the disease spectrum. These images are powerful and significant, but do not accurately represent the disorder as most individuals are considered “mildly” affected. To make matters worse, an internet query on NF1 will also uncover photos of individuals with severe symptoms completely unrelated to NF1 such as Proteus, or “Elephant Man” syndromes (Lengendre, 2011), which only adds to the confusion about what NF1 really is. Viewing these photos could misinform any patient or parent about NF1, with the greatest impact being on people who have low to moderate health literacy and are reviewing information with questionable readability, reliability and relevance.

This study aims to gather information from parents of children with a new diagnosis of NF1, who have no family history or prior knowledge of the disease about their experiences viewing internet images of people with NF1. Insights from parents about their reactions to the photographs they encountered online can help healthcare providers, including genetic counselors, adequately address common concerns about NF1 images and prepare future parents of children with NF1 for their first internet searches for NF1 information.
METHODS

Brandeis University’s Institutional Review Board reviewed and approved this study.

Study Population

We recruited study participants through the Children’s Tumor Foundation’s NF Registry (Appendix A), a confidential database of patient-entered information about an individual or child’s NF medical history. Individuals or families who are part of the registry have identified themselves as being open to research participation and have previously consented to be contacted by qualified researchers for clinical studies related to NF. We sent a recruitment notice (Appendix B) to an initial pool of 231 NF Registry members who were parents of children with a de novo case of NF1 diagnosed within the last 5 years. We encouraged potential participants to contact us directly by e-mail and those who responded to the initial recruitment notice participated in a brief screening phone call (Appendix C) to confirm they were eligible candidates. Study inclusion criteria were that individuals must: be 18 years of age or older and speak English as their primary language, be the parent of a child diagnosed with NF1 in the last five years who is the first affected person in their family, and have encountered online images of a person or people with NF1 while searching the internet for information about their child’s diagnosis.

A total of 54 individuals responded with interest to the recruitment notice, of which we screened 12 by phone and 10 were deemed eligible for study. Following the initial screening, we sent eligible candidates an informed consent document (Appendix D). We received signed consent forms from 8 participants and arranged phone interviews to be conducted at the participants’ convenience. We scheduled interviews with 7 participants; 1 individual was lost to
follow up. Each participant received a $25 Amazon gift card as a token of appreciation for their participation and donation of time.

Demographics

All participants in this study were females, who ranged in age from 30 to 40 years. The current age of participant’s children ranged from 2 to 3 years, with their reported age at diagnosis ranging from 2 to 18 months with four participants reporting confirmatory genetic testing. Participants’ affected children all had some common symptoms, including café au lait spots, skin fold freckling, Lisch nodules or neurofibromas. Four of the affected children had an optic glioma and one had tibial bowing as well. Table 1 describes further information about each participant’s child, including NF1 symptoms and other medical concerns. Table 2 describes the logistics of participant’s first internet search for information regarding NF1.

Interviews

We employed a semi-structure interview guide (Appendix E) with open-ended questions focusing on NF1 diagnostic stories, feelings about pictures participants encountered when performing internet searches, sharing of NF1 images with others, and advice for parents seeking NF1 information online. Although we used the same interview guide for each participant, the emphasis of each topic was dependent on the participant’s individual responses and issues raised during the interview. Following the audiotaped portion of the interviews, we gathered demographic information from each participant and recorded it manually. We gave participants the option to decline answering any question, or stop the interview at any time. The duration of interviews ranged from 25 to 40 minutes.

Data Management and Analysis
We audiotaped each interview using www.freeconferencecalling.com. Mulberry Studios, a professional, confidential transcription service, transcribed each digital audio recording verbatim. We de-identified all transcripts and labeled them with a unique 3 digit coded ID to protect patient privacy. We uploaded all interview audiotapes and transcripts to www.box.com, an encrypted site hosted by Brandeis University.

We analyzed demographic information manually using descriptive statistics. We employed thematic analysis using ATLAS.ti software for each interview transcript to find common themes across participant responses to questions in the four major areas of focus.
<table>
<thead>
<tr>
<th>Participant</th>
<th>Current Age of Child</th>
<th>Age of Child at Diagnosis</th>
<th>Sex of Child</th>
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<th>Less Common NF Symptoms</th>
<th>Other Medical Concerns</th>
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</thead>
<tbody>
<tr>
<td>044</td>
<td>2 years</td>
<td>4 months</td>
<td>Male</td>
<td>CAL, Freckling</td>
<td></td>
<td>Migraines</td>
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<tr>
<td>253</td>
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<td>2 months</td>
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<td>Optic Glioma, Bilateral Tibial Bowing</td>
<td>Sleep Apnea, Hole in Heart</td>
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<td>Port Wine Stain</td>
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Table 1. NF1 diagnostic information on study participants’ children
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<tr>
<td>Pediatricist</td>
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<tr>
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<td></td>
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<tr>
<td>Work cubicle</td>
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<tr>
<td>Alone</td>
<td>6</td>
<td></td>
</tr>
<tr>
<td>With husband</td>
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</table>

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<th>First Search Terms Used</th>
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<tr>
<td>Coffee spots, birthmarks, spots, café au lait spots</td>
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<td></td>
</tr>
<tr>
<td>NF, NF1, Neurofibromatosis</td>
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<tr>
<td>Genetic disorder</td>
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<td>Google Images</td>
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<td>WebMD</td>
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<td>ctf.org</td>
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Table 2. Logistics of participant’s first introduction to NF1 and first encounter with online information
RESULTS

The interview discussions focused on four main areas; diagnostic stories, reactions to NF1 images, sharing NF1 images and information, and advice for future parents of children with a new diagnosis of NF1. Initial analysis resulted in 290 codes which we used to identify 28 code families. We sorted these code families under each main category to identify key themes. Figure 1 represents the categories, key themes and sub-themes that emerged during the interviews.

Diagnostic Stories

Each participant answered a series of questions to provide an understanding of their child’s overall health and development. During this time, we asked about any of their child’s health conditions (related or unrelated to NF1), in what context they first discovered or heard the term neurofibromatosis, and what medical specialists care for their child. While hearing the diagnostic stories from these mothers, the theme of being an advocate for your child emerged.

Be an Advocate for Your Child

Four of the seven participants discovered the term “neurofibromatosis” while proactively searching the internet for health-related information concerning their child (Table 2). Of these four participants, three stressed the importance of advocating for your child, in regards to getting a diagnosis, gathering as much information as possible, and spreading awareness of NF1 to the public. Participant 253, whose pediatrician noticed multiple café au lait spots on their three year old son, stated,

“Oh gosh, I would say-- I don’t regret looking it up initially; otherwise, we never would have had a diagnosis. So I think right away, if you ever, as a parent, if you ever get the inkling that your child might have a medical issue I think it’s absolutely in your best
Participant 664 disagreed with her pediatrician’s recommendation to follow her daughter’s symptoms without an evaluation by someone familiar with NF1 or performing genetic testing. This mother had a background in genetics and pushed for her daughter to have genetic testing, which led to a definitive diagnosis. She reported feeling that doctors would have missed diagnosing her daughter’s optic glioma without this persistence. She explained,

“I referred myself to see a geneticist because I went to the pediatrician and they were like, “You know, maybe you should see a dermatologist.” We went to dermatologist that was different than the one that was seeing her for her port wine stain and told me that it was not necessary to do anything, even if she had NF, they said at that stage it was not crucial to know. And I strongly opposed because, I’m not a geneticist, but I am a molecular biologist and I have a genetics background. So I figured if they knew the mutation and this, then they could sequence it and because there could be an underlying problem since birth, then why shouldn’t we know it? […] I checked the UCLA site and then talked to my pediatrician and I wanted to have her genetically tested […] It’s a bit sad because the doctor should advise parents on what to do. It was, sorry to say, but stupid from the doctor to say, “Well, wait a few years and see,” because if we hadn’t known, then she wouldn’t be diagnosed with the optic glioma early on and if the glioma was growing, then we wouldn’t know.”

Participant 380, who was unsatisfied with her pediatrician’s initial explanation for her daughter’s café au lait spots, and by instinct knew there was a different explanation, stated,

“When she was born, I noticed that she had these spots on her and I was just told, “Oh well, it’s because she’s a mixed baby.” Because myself, I’m English, Irish, Dutch, French and Scottish. And my husband is 100 percent Mexican. So she’s a definite mixed child, or however you want to say it. And I just kept saying like, “No, there’s just something else there, there’s something else there […] So yeah, I mean it really was-- I just couldn’t-- it just wouldn’t leave my mind and I’m like, there’s something about these spots, or these whatever you want to call them, and they just won’t-- there’s just something there, you know what I mean? And she has both tibial bowing, she has double tibial bowing in her legs and it was just like there’s something there with my baby, you know what I mean?”

This mother also encouraged other future parents of children with NF1 to follow their own gut feelings and be advocates for their family as well. She said,
“So just keep the hope, but to stay strong in the sense of know what is best for your family and it’s okay to disagree with the doctor. Or not even necessarily disagree, but want a second or third opinion on something, it’s okay. And just, be an advocate, not just for NF but especially for your own child, or your own family member.”

Initial Reactions to First NF1 Images

While reviewing participant’s initial reactions to the first NF1 internet images they encountered, three key themes emerged: looking at pictures caused negative feelings, images misrepresent what NF1 is, and photographs of the most severe symptoms were the most memorable for participants.

Negative Feelings

All participants recalled experiencing negative feelings during their initial encounter with online images of individuals with NF1.

a. Worry

Each of our study participants stated that the first internet images they saw of NF1 caused them to worry for their child, and this worry stemmed from the variability of NF1 and the possibility of disfiguring symptoms. Participant 936 recalled feeling scared during her first internet search when her son was 18 months old. She explained,

“I mean, I remember being really, really scared and kind of horrified. Well, I don’t want to say that, that sounds bad. But just thinking this was something that was really deforming to this body and to the skin and that this was a pretty terrible disease and there was no treatment and no cure.”

Participant 253, who was also concerned about the disfiguring nature of symptoms she saw online, said,

“It made me scared, sad. Like I just felt scared for my son, that I didn't want my sweet baby with perfect skin to ever look like that”

Participant 380, whose daughter received a diagnosis of NF1 at two months old, added,
“Immediately. It was just like, oh my God, what is her life going to be? What's her quality of life mentally, physically, socially? You just start wondering, like oh my God, what is she in for? What will she have to endure, you know what I mean?

b. Heartbreak

Four of the seven participants used the specific term “heartbreak” when describing the images they viewed, as they felt the images depicted possible severe outcomes for their own children.

Participant 557, whose son was reportedly diagnosed at two months old, stated,

“I was just heartbroken. You know, you have a baby who’s healthy or you think is healthy, and to be as “normal” as possible, and when I saw them for the first time, like my heart just broke because I just-- initially I just thought this is what he’s going to have to deal with. I mean, not really learning about it beforehand, I just saw him and thought, “He’s not going to have a “normal” life. He's going to be different,” and I was heartbroken.”

Participant 253 described her reaction similarly, she said,

“I would say, my heart dropped out of my chest. It was just the worst feeling in the world. My baby, he was only 15 months old at the time and I just got these mental images of my baby having bumps and lumps and tumors growing all over him, all over his body and kind of disfiguring his face and things like that.”

Participant 253 also revealed her heartbreak stemmed from fearing other people would view her son as different, she explained,

“Now I think about those initial reactions and it was really very selfish. I would say selfish, that I didn’t want to go out in public and have people staring at my kid. Yeah, that would be my initial reaction.”

Participant 044 had concerns that NF1 would cause her son to be disabled, she added,

“He’s so small, it breaks your heart to know that your child is going to have any disadvantage in life.”

Participant 380, whose daughter has five NF1 symptoms, said,

“And, you know, you do what any parent does, or any person looking into something after-- you've got this little baby that is everything to you and you come across this word you can't even pronounce. And you Google and then you're like, “Oh my God, oh my God,” because when you see it with black and white words, but then you hit like images on Google, your heart just shatters. Like, the possibilities and they're endless and you
don’t know what to think and you just think the worst instantly and you realize that she will never-- even if it ends up being a mild case or not being as drastic physically that you see online, some of these pictures, you still know that it-- I mean, you're just scared. To be honest with you, we're scared every day of our lives as to what's to come for her.”

**Internet Images Misrepresent NF1**

Another major theme among participants’ reactions to the first internet images they viewed was that all felt that the majority of online pictures do not accurately reflect what NF1 is. Most participants did not feel the images were a reflection of their own child, nor did they agree that they represent the full spectrum of disease seen in the NF1 population.

*a. Images Are Not Related to Own Child’s Diagnosis*

When asked how these images relate to their child specifically, five out of the seven participants felt that the majority of NF1 pictures on the internet are not representative of their own child. Participant 577 who felt that NF1 internet images did not correspond to her son’s current symptoms and experiences, she explained,

“No. I mean, not at the point he’s at now because I know like the neurofibromas usually come, they say, around school age or puberty, if he gets them at all. I don't think necessarily it represents where he’s at right now, absolutely not.”

Participant 936 felt the images only relate to individuals severely affected by NF1, she added,

“I don’t think he’s going to be covered in head to toe. I mean, then I learned half of the cases of NF, there's no symptoms at all. People just have it and they never have symptoms. So if he doesn't have one of those cases, which is what we're hoping for, I don't think his case is going to be that bad. [...] So we're kind of able to sort of get past that and realize that he wasn’t going to be disfigured, or it doesn't look like he’s going to be disfigured by these things.”

*b. NF1 Images Are Not Representative of the Entire NF1 Population*

When asked how these images on the internet relate to all individuals with NF1 as a whole, three participants stated that internet images are not representative of all individuals with
NF1, and three other participants were unsure. Participant 253 explained that the images only represent a small proportion of individuals with NF1.

“I think my guess that this is the very-- like those images are the very worst cases of NF. And the majority of people, if it’s one in 3,000 people have NF, the majority of people don’t look anything like that.”

Participant 557 added,

“No. I mean, it might represent like a small percentage, but I don’t think it represents it as a whole.”

The three participants who were unsure if internet images they viewed represented the NF1 population and the one participant who felt that the images were representative of the NF1 population, stated that the images found on the internet accurately depicted what individual NF1 symptoms look like, but not necessarily how most people with NF1 are affected. Participant 271 felt this was difficult question to answer, she explained,

“For me, that's a hard one because I feel like NF affects everyone so differently. I mean, even just with all of like the contacts and the friends we've made here, every one of our cases is so different and so far none of them have really fit the mold of what I've seen on the internet.”

Participant 380 was the only participant who felt the images were representative of all people with NF1 as a whole, drawing upon experience with other individuals with NF1 in the community and in support groups. This mother felt that increased exposure to the NF1 population made her more aware of the ways these images are representative of NF1 individuals, she said,

“Yes. And I say that like that because I haven't run into many people with the external disformalities. […] But now being aware of what to look for and what things look like, I have run into more people. And then, of course, we've gone to the NF forum and just people in our community, if I see someone it’s like oh okay, they have NF. You start to notice things more, you know?”

Images Depicting Very Severely Affected Individuals Were Most Memorable
We asked participants to recall which image or images were most memorable from their first initial internet exploration, and all recalled a photograph depicting a very severely affected individual. Participant 380 recalled a man covered in neurofibromas, she said,

“There's one, it was of a man, and he is covered in the fibromas, exterior fibromas, the skin fibromas, and it just kind of-- it just shocked me, you know what I mean? And it was just a regular man just standing there, no big deal. And then the next one was-- I don't know if it’s the same man or not, but he was naked, total body though, from head to toe, and of course his genitals weren't blocked out or anything. But it showed a picture like half and half off the screen. The first half was the front of his body and the second half was the back of his body and he’s just completely covered in fibromas. And again, it turns into oh my gosh, my precious little perfect baby, you know? And then of course you feel guilt because you're like, “Oh my gosh, I shouldn't be speaking this way because this poor man’s dealing with it.”

Participant 044 was most struck by an image of a girl with a plexiform neurofibroma, she said,

“I remember one with a girl and she still shows up in a lot of the marketing. She has a large tumor on her face, on her jaw. She's a beautiful little girl, but she has a large, inoperable tumor on her jaw.”

Two participant recalled images of the “Elephant Man” (Figure 4). Participant 271 stated,

“Or, the Elephant Man, I don't know if you've ever heard that term....I think those were the most scary for us because we had just been told about something that we had never even heard of before and I think that photo kind of resembled the worst case scenario for everyone. And was something that patients aren’t really prepared for”

Participant 577, who also remembered seeing photos of the “Elephant Man”, commented,

“They were horrible. I remember seeing people who had tumors or growths all over their faces and their arms. I remember seeing a picture of the Elephant Man, I think that's how they referred to him. Giant tumors, out of somebody’s extremities. It's almost like you saw the worst case possible scenarios. You read more into it, and it says, “This is like worst case. Most people grow up and don’t have this.” But the pictures they show you are the worst case.”

Sharing NF1 Images and Information

During our interviews, we explored participants’ thoughts on sharing any of the images they found with other individuals, including spouses, family members, friends and healthcare providers. In discussing this topic a key theme emerged; sharing internet images of NF1 with
friends and family is a personal choice. When we asked participants specifically about sharing these images with their affected child another key theme emerged; future discussions regarding NF1 images with their children will be guided and age appropriate.

**Sharing Internet Images of NF1 with Friends and Family is a Personal Choice**

Six of the seven participants conducted their first search for NF1 images alone. Two participants chose not to share the images with any individuals, while the remaining five participants chose to share images selectively. Participant 044 recalls sharing the images with her husband, she explained,

“He cried. He cried. I mean, he's a little bit more emotional than I am, just in general, our personalities. He's more sensitive than I am. And it was really hard for him to deal with. It hit him hard. And again, that's where the conversation about the plane crash thing came out. Yeah, he had a hard time with it to the point where he said, “I can't look at this anymore.”.... For my husband, that's not the right approach. He needed time to digest and me showing him pictures or telling him what I'd learned right away while he was still feeling vulnerable from the diagnosis, or frankly even the potential for the diagnosis, didn't help the situation. He probably should have waited.”

The same mother went on to explain why she choose not to share images with anyone except her husband, she added,

“No, I don't think I shared any images with anyone. I mean, if someone had asked me, I would have sent them one, I guess. But no one asked. It never occurred to me to send someone a picture. I certainly sent them information.”

Participant 577 shared the pictures she encountered with her husband, as well as her mother, she said,

“I did share them with-- with his dad and my mom. I remember telling them about what had happened, this is before I knew what could really happen. So, yeah, I did share some of the pictures I saw and I was crying when I talked to them both and upset, yeah.....They were devastated, too, and that's pretty much it. They were trying to learn as much as they could as well and didn’t know a lot about it, yeah. So they were heartbroken and devastated as well.”
Participant 253 shared information about NF1 with family and friends, but not specific images, she stated,

“I don’t think the images. I know I’ve told people when they asked, including family. I know I’ve told people, “If you do decide to look it up, just don’t look at the images because it’s really the extreme cases and shouldn’t be what [Child’s Name] would experience.”

That same mother however, recalls sharing one specific image and story with an online support group forum. She added,

“The only thing I can remember sharing was actually after I sought out getting the genetic-- getting in with the geneticist, I joined that NF moms group on Facebook. And right around that same time is when the picture of the Pope giving the man with NF a hug came out. And I remember sharing actually the article about that on that NF moms group.”

**Future Discussions Regarding NF1 Images with Child Should be Guided and Age Appropriate**

All seven participants agreed that they would like their children to have their parental guidance when they first encounter internet images of NF1. There was also complete agreement that these conversations should be age appropriate. Participant 271 whose daughter is two and has an optic glioma, stated,

“I feel like that would spark a lot of fear in her. So finding the best way to let her know that things that you see are different and because there are different types of NF, not everyone gets a brain tumor, not everyone gets a plexiform. And so I think the internet kind of blankets things.”

Participant 577 worried about her son seeing online photos alone, she added,

“You want to protect your child. You want to protect them from things and images and for as long as you possibly can. So I would feel devastated if he saw it without somebody there helping him through the process knowing that this is just maybe worst case scenarios. This is maybe not what's going to happen to you.”

Participant 936 explained what she would plan to tell her son when he saw internet images, she said,
“I think I would say, “Well, you know, some people have this disease as more symptoms with some people than others. And you have already, when you were very small, dealt with a couple of major issues.” He’ll probably know that-- I mean, he’ll have a scar on his arm, he has a giant scar on his arm and on his leg and he still goes in every three months for an MRI and he’ll do that for the rest of his childhood. So he’ll know that that’s the extent of what he has and I think all I could do is just be like we hold that this-- it’s not likely that this is what’s going to happen to you. But if it does, we’ll treat it.”

Advice for Future Parents of Children with a New Diagnosis of NF1

Finally, we asked participants what advice they would give to parents of children with a new diagnosis of NF1 who were seeking information online. All seven participants recommended that parents seeking NF1 related information visit the Children’s Tumor Foundation website. Two key themes emerged from these discussions; parents should rely on trustworthy sources of information and support groups, and they should be aware that most people with NF1 do not have the disorder as severely as the individuals depicted online.

Rely on Trustworthy Sources of Information and Support Groups

All seven participants encouraged future parents to seek out accurate information from sources such as healthcare providers, reputable websites, and other individuals with experience with NF1. Participant 253 warned against “Google” and said,

“I would look for reputable sources, don’t just Google image things like I did.”

Participant 044 advised parents to only use internet material as supplemental information, relying primarily on recommendations from healthcare providers. She explained,

“Okay. So for information, my first advice would be go to a reputable source. Sign in to Google and look up what Wikipedia has to say and Web M.D. and all those other sites, but speak to your doctors first about what they see already with your child. And see what sites the experts in the medical field recommend have the most accurate information and start there.”

Participant 271 felt that seeking information from individuals who have experience with NF1 is most valuable, she explained,
“I think the best thing to do is to talk to people who have had someone diagnosed with NF that kind of know, or to wait to see your geneticist or your specialist that can kind of guide you into an area where you can find honest and accurate information.”

**Be Aware That Most People with NF1 Are Not as Severely Affected as Individuals Depicted Online**

All seven participants agreed that the images of individuals with NF1 online depict the “worst case scenario” of NF1 and felt that future parents dealing with a new diagnosis of NF1 in a child need to know this. Each participant felt that ratio of severely to mildly affected individuals found in internet photographs did not reflect the actual ratio of severely to mildly affected individuals in the NF1 population. Participant 936 explained,

“Just that what they’re going to see are very extreme cases and that before they start making assumptions about what their kid is going to look like or have or be like, they should talk to an NF specialist and also just wait and see before they start getting really scared about what it’s going to be like. Because like I said, we learned there’s a whole range of severity with this thing and so don’t jump to conclusions based on what you see on the internet and talk to someone who really, really knows this disease.”

Participant 577 suggested that parents should look at written materials before viewing the pictures, as the images do not accurately portray most people with NF1. She advised,

“Yeah. Just to get as much written information as they can from as many places as they can before they view the pictures because the pictures-- yeah. They paint a different picture, maybe, than what your child may go through. And just to get as much information from other sources than images.”

Participant 253 added,

“And just know, keep in the back of your mind, that the majority of people with NF don’t develop these extreme disfiguring appearances.”

Participant 044 felt the images from an NF1 search depicting severe symptoms or deformities may not even be of individuals with NF1, she said,

“I mean, there's images that come up that I'm not even really sure are related to NF with severe deformities”
Figure 1. Emergent Categories, Themes and Sub-Themes
DISCUSSION

The seven qualitative interviews with mothers of children with a new diagnosis of NF1 conducted as part of this study provided valuable insights into the impact of internet images of NF1 on parents of newly diagnosed children who had no prior knowledge of the condition.

Diagnostic Stories

The major theme that emerged while discussing diagnostic stories with participants was the importance of being an advocate for your child. All of the mothers valued being proactive in their child’s care, and felt an important avenue for this was exploring potential diagnoses themselves and becoming educated about NF1. This often resulted in participants seeking health related information on the internet. While this is in keeping with previous studies demonstrating a high incidence of online exploration by parents of children with genetic conditions (Roche, 2009), their queries were unique in that many were searching for primary information about concerns in their child. In fact, as described in Table 3, four of the seven participants discovered the term “neurofibromatosis” themselves while conducting research online using search terms that described their child’s symptoms, most commonly “café au lait spots”. Of the four who discovered this term independently, three felt like they would not have a diagnosis for their child without being proactive and seeking out information by their own accord. Participant 664 strongly agreed with this, explaining that pushing for genetic testing with her pediatrician and a self-referral to a geneticist was the reason her daughter received an early diagnosis which subsequently led to investigations uncovering an optic glioma. The mothers in our study
demonstrated that, in certain situations, being proactive and looking for information on the internet can be instrumental in getting the best care for your child.

*Initial Reactions to First NF1 Images Viewed Online*

The three themes that emerged when discussing reactions to the first encountered NF1 images were negative feelings, online images misrepresenting NF1 and very severe manifestations being the most memorable images. All participants expressed that their first reactions to NF1 internet images were negative feelings. The most common feelings participants remember having were worry for their child or heartbreak. Every mother was concerned for their child’s future based on the severity of symptoms they encountered in online images and the inability to predict what complications might occur in them. The apprehension about development of severe symptoms tended to be more prevalent among parents of children who only had common NF1 symptoms, such as café au lait spots and freckling. This could be because these findings are benign and require no medical monitoring, allowing these children to not really appear “different”, yet because other signs are age dependent, they are not guaranteed to be mildly affected their entire life. Furthermore, parents of children with less common, more medically significant, symptoms may come to terms with the fact that their child may develop complications reflected in the online images sooner than parents of children with minimal visible differences. Therefore, parents whose children are basically healthy despite their diagnosis of NF1, might be most frightened at the images they see online, and require guidance or counseling from their healthcare providers on how these may not accurately reflect the disorder.

Another common negative feeling was heartbreak, as the internet images gave more tangible meaning to the diagnosis beyond written information. The mothers in this study reported feeling sad that their child could potentially suffer both physically due to NF1 health
complications, and psychologically due to the stigma associated with having a severe visible difference or disability. Of the four participants who expressed heartbreak, three explained this feeling stemmed from the concern that people would treat their child differently if they developed disfiguring symptoms like the individuals in the online images. They stated that they would not want their child to be disadvantaged or labeled as different based on their diagnosis. This feeling could relate again to the need parents feel to protect and preserve their child’s well-being or reflect the realization that their children may lead different lives than their mothers had envisioned. In any case, it would be helpful to prepare parents of newly diagnosed children for these strong feelings and provide resources to help them cope with their emotions.

Thoughts about the representative nature of online images varied between participants but many felt, as a whole, they misrepresented what NF1 really is. Five of the seven mothers we interviewed specifically felt that the majority of internet images did not relate to their own child’s experiences with NF1. This is likely because many of the participants’ children are young and did not have visible difference beyond café au lait spots and freckling (Table 1). However, two participants reported that a few images related specifically to some of their child’s symptoms. These women explained that, although their children may not have many severe, visible differences such as those depicted in the photographs, they do have some symptoms in common with the individuals depicted in the online images. For example, Participant 380 explained that her daughter has a neurofibroma on her scalp, which resembles neurofibromas depicted in online images. So, although her daughter has this symptom, this mother realizes that she is not affected in the same way as individuals pictured who have neurofibromas covering their entire body. Parents of young children with NF1 need to be able to put the information they
see on the internet in perspective with regard to their own child, and this may require guidance from either healthcare providers or others who have experience with NF1.

The course of NF1 is unpredictable and the presentation of symptoms is extremely variable between affected individuals. However, parents of children with a de novo mutation and no prior knowledge of NF1, like our participants, would not know this fact. None of the mothers in our study knew anyone with NF1 except for their own child prior to their child’s diagnosis. Therefore, it would be reasonable to think they would assume online images represent the course of the condition for the entire NF1 population. Yet, three participants stated they felt that the online images they viewed do not reflect the NF1 population as a whole and three participants expressed that they were unsure. The participants who were unsure if the images reflected the NF1 population thought it was difficult to determine due to their lack of exposure to other individuals with NF1 at that time. They did feel though, that the pictures they viewed accurately depicted specific symptoms, just not the overall impact of NF1 on the average affected individual. Learning more about NF1 through their child’s healthcare providers, reputable internet sources, and exposure to the NF1 community, allowed participants to eventually come to different conclusions about the representative nature of internet images for the entire NF1 population. All participants expressed that they know now that images they initially encountered did not represent the full NF1 spectrum and they are now able to better understand how what they see on the internet actually relates both to their child and to NF1 as a whole.

When asked what stood out the most from their first encounter with online NF1 images, all participants described images of individuals with severe, disfiguring symptoms. The most common memorable images included a man who had neurofibromas covering his entire body (Figure 2), individuals with disfiguring plexiform neurofibromas (Figure 3), and images of the
“Elephant Man”, Joseph Merrick (Figure 4). Figures 2-4 are not the actual pictures seen by participants, but are representations of the most common images described by participants. For all of the mothers in our study, these images represented the worst-case scenario of how NF1 could potentially affect their child. However, most participants were not aware that these images were only representative of a small portion of individuals with NF1 until they gathered more information following their first image search. Participants described these images as memorable because they were the most shocking. Three participants cited images of the “Elephant Man” as memorable, however only 1 of them clarified that they later learned the “Elephant Man” did not have NF1. This has been a well-documented misconception about NF1 that surprisingly still persists even in some individuals who are closely involved with the NF1 community, and as a result, the “Elephant Man” still represents the most severe form of NF1 for some people (Legendre, 2011). It is important for parents exploring information on NF1 for the first time to know that the “Elephant Man” did not actually have NF1 and that the photos of him have nothing to do with this disorder.

Sharing NF1 Information and Images

All participants were open to sharing their child’s diagnosis, however many expressed that choices about sharing NF1 information and images were very personal and individual to each situation. Mothers who did choose to share the images they encountered on the internet with others were very selective, only sharing with spouses and/or very close family and friends. They feared that sharing images with friends and family would cause them to experience the same negative feelings they themselves had when looking at online information for the first time. This is not surprising, given that other family members or friends likely have less knowledge of what NF1 is, and therefore are more apt to fear the worst when seeing information that does not
present a balanced view of the disease spectrum. It is also possible that the hesitation to share NF1 information and images relates to the worry participants felt about people treating their children differently, or viewing them as disabled due to their diagnosis.

None of our study participants reported sharing internet images of NF1 with their children at this time. These mothers stated that the reason for not sharing these images is that their child is too young, which is not surprising given that all of the participants’ children were three years of age or younger. They went on to explain that they felt children at this age were too young to understand that although they share the same diagnosis as the individuals in the images, they have a different presentation of the disease. However, all did express interest in having guided conversations with their child about how internet images depict NF1 when their children were old enough to understand the diagnosis better. The most common thing parents said they would want their children to know is that images found online represent the most severe cases of NF1. The participants agreed that that they hope their child never encounters the images independently, as they might worry about their own futures in regards to NF1.

Advice for Future Parents of Children with a New NF1 Diagnosis

When asked what advice participants would give to parents who had a child newly diagnosed with NF1, all recommended using trustworthy sources of information, including the CTF website. Given that we recruited participants from the CTF NF Registry, it is not surprising that they were more likely to favor this website. At the same time, all of the mothers we interviewed encouraged future parents to seek information from other reputable internet sources as well. One woman specifically would warn future parents to avoid searching for information using Google, as the wealth of information can be overwhelming and not all information is accurate or reliable. Furthermore, image searches on Google bring up pictures without
descriptions or explanations, which do not provide families with the most accurate portrayal of what NF1 is. Most participants expressed that it is inevitable that human curiosity will cause parents to seek information about their child’s health and well-being from the internet. However, all stressed that the most valuable and trustworthy information comes from healthcare professionals and support groups made up of people who have experience with NF1.

All of the mothers in this study also emphasized that parents seeking information about NF1 need to know that the images of affected individuals found on the internet are not balanced. The consensus among participants was that the number of images on the internet depicting severely affected individuals is not in proportion with the number of individuals with NF1 who actually develop severe symptoms. Therefore, seeing internet images may mislead parents with little information about the severity of the disorder and cause them to fear the worst for their child when, in fact, many individuals with NF1 live relatively healthy lives. Most participants expressed that realizing the online pictures do not present a balanced view of NF1 helped them to cope with their reactions to the images later on. Mothers in this study stated that they learned that most people with NF1 do not have severe symptoms by speaking with healthcare providers and other families affected by NF1 involved in support groups, and encouraged other parents of newly diagnosed children to do the same.

Limitations

This was an exploratory qualitative study designed to explore the impact of NF1 internet images on parents of children recently diagnosed with NF1 and no prior knowledge of this condition. There were only seven participants who were all were mothers, and six of the seven were Caucasian. And, while study participants were from a range of geographic areas within the
United States, the perspectives of this small sample size might not be reflective of the larger population of parents with young, newly diagnosed children.

We recruited participants online through the CTF NF Registry, which may have introduced sample bias. It is possible that individuals involved in support organizations, like CTF, have children with more severe NF1 and might be more inclined to participate in the CTF NF Registry and be open to research opportunities. In fact, four of the seven participants had children with less common NF1 symptoms, including optic glioma, two of which were progressive, and tibial bowing. The participants in this study had children diagnosed at very young ages, all before the age of 2 years. While children may come to the attention for suspicion of NF1 at early ages, few children receive a diagnosis at less than 6 months with no family history of the disorder. Therefore, the children of the participants in this study may not be representative of the spectrum of disease features in young children with NF1.

Finally, this study employed qualitative data analysis, and therefore the author subjectively created all emergent codes and themes.

Future Research

The current study revealed valuable insights into the impact of online images of NF1. The use of qualitative techniques enabled us to identify the main issues for families who choose to conduct an image search while exploring the possibility of, or coping with a new diagnosis of NF1. The themes described in this study can guide future qualitative or quantitative research on this topic.

Future studies should include a larger sample size, including perspectives from fathers and parents of older children and should also distinguish between parents of severely affected children and parents of mildly affected children. It would be interesting to explore how a
family’s personal experience with the disease influences their views on the representative nature of internet images of NF1. As many children do not receive a diagnosis for several years after the clinicians raise the initial suspicion, a longitudinal study to follow families and track how their view of internet images of NF1 changes over time as their child does or does not develop new NF1 symptoms could be meaningful. In addition, since all study participants reported using the internet frequently for personal and professional purposes, it would be worthwhile exploring the reactions from NF1 image exposure in parents who have less experience conducting online research. Finally, given that prior studies suggest health literacy influences the way a person interprets online information (Gutierrez, 2014; Mcinne, 2011), future studies should assess health literacy and evaluate its influence on parents’ perception of NF1 internet images.
CONCLUSION

This study explored the impact of NF1 internet images on mothers of children with a new diagnosis of NF1 and no prior knowledge of the condition. Participants in this study expressed that the most memorable images reflected severe cases of NF1 and caused negative feelings such as worry and heartbreak. Participants stated that sharing NF1 images and information was a personal decision, and all were selective in sharing with close family or friends after they first encountered the images. Every mother in our study felt that their children should not encounter NF1 images without guidance, and conversations between themselves and their children should occur in an age appropriate manner. Participants thought that the images online misrepresent NF1, however some were still unsure whether the online images accurately represented the NF1 population as a whole. Advice for future parents of children with NF1 included using reputable sources for information and realizing that images online depict the most severe end of the NF1 spectrum. Healthcare providers, especially genetic counselors, should be aware of the impact of these images on parents of children with a new diagnosis or suspicion of a diagnosis. By starting conversations about these images early, healthcare providers can clarify misconceptions and anxieties about the likelihood of a child developing severe symptoms may be eased.
Figure 2. Man covered in neurofibromas. From Google Image search- “neurofibromatosis”

Figure 3. Plexiform neurofibroma, From Google Image search- “neurofibromatosis”
Figure 4. Joseph Merrick, the “Elephant Man”
BIBLIOGRAPHY/CITATIONS

Mary-Kate Cushing
Brandeis University
Genetic Counseling Program
November 26, 2014

Ms. Cushing:

You have permission from the Children’s Tumor Foundation (CTF) to use our NF Registry to recruit subjects for your study, “The Impact of Online Images of People with Neurofibromatosis Type 1 (NF1) on the Parents of Newly Diagnosed Children.” The CTF administrators responsible for the Registry have reviewed your study documents and approved your study for this use.

The NF Registry is a confidential dataset of patient-entered information about an individual’s, or their child’s, NF medical history. Its main purpose is to provide de-identified data to qualified researchers and to speed recruitment for clinical studies in NF. The Registry helps recruitment by sending emails to registrants who have previously consented to such contact. Emails can be sent to a targeted audience within the Registry based on searchable characteristics. In the case of minors, the parent or guardian registers for the child(ren) and is the point of contact for all communications with the Registry administrators at CTF.

Recruitment text is supplied by the researcher. It describes the research and provides contact information for the registrant to contact the researcher. For the present study, the Registry, which contains 4,500 patient accounts, can offer an initial recruitment pool of 962 parents of children with de novo cases of NF1.

Pamela Knight, M.S.
Clinical Program Manager
Children’s Tumor Foundation
120 Wall St.
New York, NY 10005
IMPACT OF ONLINE IMAGES OF PEOPLE WITH NEUROFIBROMATOSIS TYPE 1 (NF1)

My name is Mary-Kate Cushing and I am a graduate student in the Genetic Counseling Program at Brandeis University. I am working on a research project to explore the impact of online images of people with NF1 on the parents of newly diagnosed children who are the first affected person in their family. I am in search of volunteers willing to participate in a qualitative study in which they will discuss their experiences conducting internet research for information about NF1 and encountered online images of a person or people with NF1 there. We hope that the information gathered from this study will help genetic counselors, healthcare professionals, as well as other families navigate the online search for NF1 information and understand the implications of NF1 online images.

Participation in this study is voluntary and open to parents who:

- Are 18 years of age or older
- Speak English as their primary language
- Parents of a child with a clinical diagnosis of NF1 who:
  - Is the first person affected in their family (a de novo or new mutation)
  - Was diagnosed within the last five years
- Parents who encountered online images of a person or people with NF1 while searching the internet for information about their child’s diagnosis

Potential participants will be asked to partake in a brief phone interview to assess eligibility for this study. Following this conversation, eligible participants will be asked to participate in a 30-45 minute audiotaped telephone interview at their convenience. As we appreciate participants donating their time and insights, all participants will be given a $25 gift card to Amazon.com. I hope to have all interviews complete by March 6, 2015.

To protect participant privacy, all identifying information of participants will be kept confidential and will be destroyed after completion of the study. Identifying details will be changed to protect the privacy of the participants.

If you are interested in participating in this study, please contact Mary-Kate Cushing by email at mcushing@brandeis.edu by February 28, 2015.

I appreciate your interest in this study and look forward to hearing from you.

Sincerely,

Mary-Kate Cushing
Genetic Counseling Graduate Student
Brandeis University
Waltham, MA
THE IMPACT OF ONLINE IMAGES OF PEOPLE WITH NEUROFIBROMATOSIS TYPE 1 (NF1) ON THE PARENTS OF NEWLY DIAGNOSED CHILDREN

Demographics and Eligibility Screening:

Name: ___________________ Age: _____ Gender: ______________

Phone Number: (____) ______-__________ Email: ___________________

City and State you live in: _____________________________

[Symptomatic]Child’s Name: _________________ Child’s Age: ________

Age at diagnosis: _____

Son ☐ Daughter ☐

Do you have other family members with NF?

Yes ☐ No ☐

Did you have any knowledge or experience with NF prior to your child’s diagnosis?

Yes ☐ No ☐

Can you tell me about your child’s NF symptoms?

Café au Lait macule(s) (“brown spots”) ______
Neurofibroma(s) (“lumps or bumps on the skin”) ______
Skin ______
         Plexiform ______
Lisch Nodules (“eye findings, benign spots on iris”) ______
Freckling ______
         Axillary (“armpits”) ______
         Ingual (“groin”) ______
Optic Glioma ______
Osseus Lesion (“problems with bones”) ______
Other NF associated medical issues______
Does your child receive special services at school or have a learning disability?

Yes ☐  No ☐

Have you conducted an internet search seeking information about your child’s NF diagnosis?

Yes ☐  No ☐

Have you encountered images of a person of people with NF online?

Yes ☐  No ☐

Are you willing to be audiotaped during the interview that will be conducted for this study?

Yes ☐  No ☐
INTRODUCTION

Gretchen Schneider is the Co-Director of the Clinical Training for the Brandeis University Master’s Program in Genetic Counseling Program. Mary-Kate Cushing is a Masters candidate in the Genetic Counseling Program at Brandeis University. This is a research study exploring the experiences of parents of children with a clinical diagnosis of neurofibromatosis type 1 (NF1) and no family history of NF1 who have conducted internet research and encountered online images of a person or people with NF1.

You are being invited to participate in this study because you are a parent of a child with a clinical diagnosis of NF1.

Taking part in this research study is completely your choice. You should not feel any pressure to participate. You can choose to stop taking part in this research study at any time, for any reason.

Please read all of the following information carefully. Ask any questions that you have about this research study. Do not sign this consent form unless you understand the information in it and have had your questions answered to your satisfaction.

If you decide to take part in this research study, you will be asked to sign this consent form. You will be given a copy of the signed form, which you should keep for your records. It has information, such as important names and telephone numbers, to which you may wish to refer in the future.

PURPOSE OF STUDY

The purpose of this study is to explore the experiences of parents of children with a new diagnosis of NF1 who have encountered online images of a person or people with NF1. Specifically, we would like to explore the experiences of parents of an affected child with no family history of NF1 and little to no knowledge of NF1 prior to their child’s diagnosis. We plan
to investigate the circumstances surrounding the parents’ first internet search for NF1 information and/or images, the impressions made by these images on parents, and how parents chose to share, or not share the images they encountered with other individuals, family members and their affected child. We hope that the experiences shared by the parents participating in this study will provide valuable insight for parents in similar situations in the future, genetic counselors and other health care professionals.

PROCEDURES TO BE FOLLOWED

You will be asked to participate in a 30 to 45 minute audiotaped phone interview. During this interview, you will be asked questions regarding your experience with online images of a person or people with NF1. The interview will be audiotaped, transcribed, and studied.

RISKS

Participation in this study presents no more than minimal risk.

However, it is possible that taking part in the interview could cause distressing thoughts and feelings. Should that occur, Sara Ellingwood is available to provide additional support.

Sara Ellingwood, MS, CGC
Certified Genetic Counselor
Maine Medical Partners, Pediatric Specialty Care
207-662-5522 x8
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There is a risk of loss of privacy or confidentiality if participants identifying information was revealed.

BENEFITS

There will be no direct benefit to you for your participation in the study. We hope that in the future, information obtained from this study will help us gain a better understanding of the experiences of parents conducting independent, online research for NF1 information and/or images.

ALTERNATIVE

An alternative is to not participate in this research study.

PRIVACY AND CONFIDENTIALITY

All records containing identifying information, such as names, email addresses, telephone numbers, and home or work addresses will be kept strictly confidential during the study. All study related documents and materials (including eligibility questionnaires, interview transcripts and audiotapes) will be kept in a secure location accessible only to the Principal Investigator and student researcher, and any databases containing identifiers will be password protected using a password known only to the Principal Investigator and student researcher. Transcripts, interview notes, and audiotapes will be labeled with a coded ID number, which will be assigned to you
upon enrollment into the study. If you are quoted or referred to in any written or oral reports of the study, you will be given an alternate name. You will never be referred to by your real name or any other identifying information in any written or oral reports based on the interview.

**PAYMENT**

As we are appreciative of participants’ time, you will receive a $25 gift certificate to Amazon.com for participation in the research study. Participants who choose to discontinue the interview early, or skip questions during the interview will still receive the $25 gift certificate.

**COST**

There will be no cost to you to participate in the study, other than the time it takes to conduct this interview.

**WHOM TO CONTACT**

If you encounter any problems related to study participation or have questions about the study, you may contact the student researcher, Mary-Kate Cushing, by email at mcushing@brandeis.edu, or by telephone at (774)313-6571.

You may also contact the Principal Investigator for this project, Gretchen Schneider, at gretchen@brandeis.edu.

If you have questions about your rights as a research study subject, contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8133.
PARTICIPANT’S STATEMENT

I have read this consent form and have discussed with Mary-Kate Cushing the procedures described above. I have been given the opportunity to ask questions, which have been answered to my satisfaction. I understand that any questions that I might have will be answered verbally or, if I prefer, with a written statement.

I understand that my participation is voluntary. I understand that I may refuse to participate in this study. I also understand that if, for any reason, I wish to discontinue participation in this study at any time, I will be free to do so.

If I have any questions concerning my rights as a research subject in this study, I may contact the Brandeis Committee for Protection of Human Subjects by email at irb@@brandeis.edu, or by phone at 781-736-8133.

I have been fully informed of the above-described study with its risks and benefits, and I hereby consent to the procedures set forth above.

I understand that as a participant in this study my identity and data relating to this research study will be kept confidential.

I agree to be audiotaped during the interview portion of this study.

YES □ NO □

__________________________  ___________________________
Date                    Participant’s Signature

__________________________  ___________________________
Date                    Student Researcher Signature
THE IMPACT OF ONLINE IMAGES OF PEOPLE WITH NEUROFIBROMATOSIS TYPE 1 (NF1) ON THE PARENTS OF NEWLY DIAGNOSED CHILDREN

Below is a general guide that we will use to lead our individual interviews. We may modify this guide as needed.

Participant Name: ___________________

Date: ___________

Time: ___________

Introduction

Thank you for agreeing to participate in this interview to discuss your experience with images of neurofibromatosis type 1 (NF1) found on the internet. As the parent of a child with NF1, your insights on this topic are valuable and we appreciate you sharing your experiences with us. We hope that the information gathered from these interviews can help genetic counselors, healthcare professionals, as well as other families navigate NF1 internet research and understand the implications of these internet images.

This interview should take about 30-45 minutes to complete. Is now still a good time to complete the interview?

You may stop the interview at any time if you feel uncomfortable proceeding. You may also choose to skip questions. If you do choose to skip a question or stop the interview, you will still receive the Amazon gift card.

This interview will be audiotaped and transcribed by a confidential, professional transcription service. This is so I won’t miss anything you say. Before data entry, your name as well as any potential identifying information that could link the transcription to you will be removed.

Before I begin the interview, I would like to ask if you have any questions about the informed consent form I sent to you. Was the form clear? Do you have any questions about it? Do you voluntarily consent to participating in this study? Do you have any other questions before I begin the audio-taping?
I. **Warm Up:** To begin, I would like to ask you some general, personal questions about your family, your child and your experiences with NF1.

a. Can you tell me about your child?
   i. Current age and grade in school (if applicable)
   ii. Personality, interests
   iii. Role in the family
   iv. General health
      1. Any medical issues?
      2. Any school/learning issues?
      3. Seen by a specialist for reasons related or unrelated to NF?

b. When was the first time you heard the term neurofibromatosis?
   i. Who did you hear it from?
   ii. In what context did they bring it up (i.e. your child has some brown spots vs. your child has NF)
   iii. Did they refer you to see a specialist (like a geneticist, genetic counselor or NF clinic)
      1. If, so who did they refer you to?
        a. How long did it take for you to get an appointment with them?
        b. How many times have you been seen by that specialist?

II. **Internet Searching on NF:** I would like to ask you some questions regarding your typical Internet use, as well as the circumstances and experiences surrounding the first time you encountered NF images on the Internet.

a. How do you use the Internet?
   i. Do you use a computer frequently? (for work, personal use, etc.)
   ii. Do you use the Internet to seek out information regularly on the Internet? (i.e. do you Google everything?)
   iii. What do you use the Internet for?
      1. Do you use the internet to find health information?

b. Can you describe to me the first Internet images you saw of a person or people with NF?
   i. What features did the photos depict?
   ii. Were they of adults or children?
iii. What Internet sites were they from?
iv. Were they medical images or pictures or people from a support group, foundation, or other organization?
v. Did you search for additional images? Why or why not?
c. Can you tell me about the first time you searched for information about NF on the Internet?
   i. Where were you? (at home, at work, in a public place i.e., a library)
   ii. Was this on a computer or smart phone?
   iii. Were you by yourself (or with a partner, friend, co-worker, etc.)
   iv. When was this (after ped brought up the dx, after seeing a specialist, etc)
   v. Why did you choose to search the Internet?
   vi. What were you looking for? (information, pictures, other families)
   vii. What search terms did you use and why?
   viii. Did you specifically search for images? Why or why not?

III. Feelings about NF Images: I would like to know more about your initial reactions and feelings to the first images of NF you saw on the internet.

   a. Can you describe to me how the images you saw on the Internet made you feel?
      i. For any emotion can you explain why you felt that way?
         1. Shocked, anxious, afraid, upset, angry, confused
      ii. Do any particularly good or bad images stand out to you?
         1. If so, what were they and what was good or bad?

   b. How do you feel these images relate to your child’s experiences with NF?
      i. Do you feel these images are representative of your child? Why or why not?

   c. How do you feel these images relate to most people with NF?
      i. Do you feel the images you have seen on the Internet are representative of most people with NF? Why or why not?

IV. Sharing of NF Information: Some families choose to share information they find on the internet with others, while some families choose not to share. I would like to know more about your experiences in making the decision of whether to share or not to share the images you have encountered on the internet of NF.

   a. Did you choose to share the images you found online with any individuals outside of your family (teachers, friends, neighbors, co-workers, etc.)?
      i. If yes,
1. Who did you share these with?
2. Why did you choose to share with them?
3. How did you share them (send them a link, show them yourself, etc.)
4. Was this the first time they had ever heard of NF and/or were these the first images they had seen?
5. Did you talk with them about these images after they had viewed them? If yes, can you tell me how that conversation went?
6. What were their reactions to these images?

ii. *If no,*
1. Why did you choose not to share these images with family members?

iii. Specifically, did you talk with any **healthcare professionals** about the images you saw on the internet? If yes, can you tell me about that conversation?
   1. Did you find talking about these images with a health care professional helpful?
   2. Did any healthcare professional specifically tell you to avoid searching the internet for information or images?

b. Did you choose to share the images you found online with **other family members**?

i. *If yes,*
   1. Who did you share these with?
   2. Why did you choose to share with them?
   3. How did you share them (send them a link, show them yourself, etc.)
   4. Were these the first images they had seen or had they found images on their own?
   5. Did you talk with them about these images after they had viewed them?
      a. If yes, can you tell me how that conversation went?
   6. What were their reactions to these images?

ii. *If yes, multiple family members*
   1. Were these conversations or reactions different between the family members?
      a. How did the conversations/reactions differ?
      b. Of these conversations, which was the most comfortable? Why?
c. Of these conversations, which was the least comfortable? Why?

iii. If no,
1. How did you come to this decision?

c. Did you choose to share the images you found online with your child?

i. If yes,
1. How did you come to that decision?
2. Did you focus on any particular images?
3. Did you exclude any particular images?
4. How did you share them (send them a link, show them yourself, etc.)?
5. Were these the first images they had seen or had they found images on their own?
6. Did you talk with them about these images after they had viewed them?
   a. If yes, can you tell me how that conversation went?
7. What were their reactions to these images?

ii. If no,
1. How did you come to that decision?
2. Do you plan to share these images with them at another time? If so, when?
3. What would you do if your child came upon images without your guidance?
   a. How would you feel about that?
   b. How would you respond?

V. Advice for Parents Looking for Internet Information

a. What advice would you give to parents seeking information about NF?
   i. Specifically, what advice would you give to parents seeking this information on the internet?

b. What sources of information would you tell parents to avoid?
   i. Are there any specific websites that come to mind?

c. What sources of information would you tell parents to seek out?
   i. Are there any specific websites that come to mind?
d. What do you think is the best time for a parent to seek out information about NF?
   i. Before referral to geneticists, after speaking to specialist, upon new symptoms arising, etc.

Before I end the audio-taping, is there anything you wish I had asked about but did not during this interview?

End Audio-Taping

Demographic Information

Age: ______

Ethnicity:
  o White
  o Hispanic
  o African American
  o Native American
  o Asian
  o Pacific Islander
  o Other

Marital Status:
  o Single
  o Married/Domestic Partnership
  o Divorced
  o Widowed
  o Separated

# of Children: ______

Geographic Region:
  o Midwest - IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, SD, WI
  o Northeast - CT, DC, DE, MA, MD, ME, NH, NJ, NY, PA, RI, VT
  o Southeast - AL, AR, FL, GA, KY, LA, MS, NC, SC, TN, VA, WV
  o Southwest - AZ, NM, OK, TX
  o West - AK, CA, CO, HI, ID, MT, NV, OR, UT, WA, WY
Highest level of education completed:
- Elementary school
- Completed some high school
- High school graduate
- Associate degree
- Bachelor's degree
- Master's degree
- Ph.D., law or medical degree
- Other advanced degree beyond a Master's degree

Employment Status:
- Employed Occupation: ________________
- Out of work
- Homemaker
- Student
- Military
- Retired
- Disabled

This concludes the interview. How was the interview experience for you?

I would like to thank you for your time and for participating in this interview. Your answers provided valuable insights to this topic. It is my hope that the information gathered from this study will help other parents and families in similar situations interpret Internet images of NF1 in the future.

I would like to remind you that your answers will remain confidential.

As a token of appreciation for your time, I will be sending you a $25 Amazon gift card via email later today.

As part of my Masters Thesis requirement for the Brandeis Genetic Counseling program I will be presenting the results of this study in May of 2015. If you are interested in knowing the results of the study please contact me via email (mcushing@brandeis.edu) and I will send them to you at that time.